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(54) Title: PRENATAL DIAGNOSIS USING CELL-FREE FETAL DNA IN AMNIOTIC FLUID

(57) Abstract: The present invention relates to improved methods of prenatal diagnosis, screening, monitoring and/or testing. The inventive methods include the analysis by array-based hybridization of cell-free fetal DNA isolated from amniotic fluid. In addition to allowing the prenatal diagnosis of a variety of diseases and conditions, and the assessment of fetal characteristics such as fetal sex and chromosomal abnormalities, the new inventive methods provide substantially more information about the fetal genome in less time than it takes to perform a conventional metaphase karyotype analysis. In particular, the enhanced molecular karyotype methods provided by the present invention allow the detection of chromosomal aberrations that are not often detected prenatally such as microdeletions, microduplications and subtelomeric rearrangements.



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